

**Table 2. GFAP Mutations Identified in Individuals with Alexander Disease**

Exon	Nucleotide Change (amino acid change)	Infantile	Juvenile	Adult	Total Affected Individuals	References
1	187A $\rightarrow$ C (K63Q)			1	1	Li et al 2005 van der Knaap et al 2005
1	208C $\rightarrow$ T (R70W)			1	1	Salvi et al 2005
1	218T $\rightarrow$ G (M73R)		1		1	Gorospe et al 2002
1	218T $\rightarrow$ C (M73T)	1			1	Li et al 2005
1	226C $\rightarrow$ T (L76F)	2			2	Rodriguez et al 2001 Li et al 2005
1	226C $\rightarrow$ G (L76V)	1			1	Li et al 2005
1	229A $\rightarrow$ T (N77Y)	1			1	Rodriguez et al 2001
1	230A $\rightarrow$ G (N77S)	1			1	Li et al 2005
1	234C $\rightarrow$ A (D78E)		1 <sup>1</sup>	5 <sup>1</sup>	6	Stumpf et al 2003
1	235C $\rightarrow$ G (R79G)	1			1	Gorospe et al 2002
1	235C $\rightarrow$ T (R79C)	8	2		10	Brenner et al 2001 Gorospe et al 2002 Shiroma et al 2003 Probst et al 2003 Ma et al 2005 Li et al 2005
1	236G $\rightarrow$ A (R79H)	11 <sup>2</sup>	2		13	Brenner et al 2001 Rodriguez et al 2001 Gorospe et al 2002 Meins et al 2002 Brockmann, Meins et al 2003 Asahina et al 2006
1	236G $\rightarrow$ T (R79L)	1			1	Shiroma et al 2003
1	256_259delinsGAGT (K86_V87delinsQF)		1		1	van der Knaap et al 2006
1	260T $\rightarrow$ G (V87G)			3 <sup>3</sup>	3	Okamoto et al 2002

Exon	Nucleotide Change (amino acid change)	Infantile	Juvenile	Adult	Total Affected Individuals	References
1	262C→T <b>(R88C)</b>	3	7		10	Rodriguez et al 2001 Gorospe et al 2002 Guthrie et al 2003 <sup>4</sup> Nobuhara et al 2004 Li et al 2005 Kyllerman et al 2005 van der Knaap et al 2006
1	262C→A <b>(R88S)</b>	1			1	Rodriguez et al 2001
1	269T→C <b>(L90P)</b>	1			1	Suzuki et al 2004
1	278A→C <b>(Q93P)</b>		1		1	Kyllerman et al 2005
1	290T→C <b>(L97P)</b>	2			2	Meins et al 2002 Li et al 2005
1	380_385dupGCGGCT <b>(R126_L127dup)</b>		1			van der Knaap et al 2006
4	619G→A <b>(E207K)</b>		1		1	Li et al 2005 van der Knaap et al 2005
4	619G→C <b>(E207Q)</b>		1		1	Li et al 2005 van der Knaap et al 2005
4	628G→A <b>(E210K)</b>		1	2	3	Li et al 2005 Kyllerman et al 2005 van der Knaap et al 2006
4	667G→C <b>(E223Q)</b>			1 <sup>5</sup>	1	Brockmann, Dechent et al 2003
4	704T→C <b>(L235P)</b>		3 <sup>6</sup>		3	Li et al 2005 van der Knaap et al 2005
4	715C→T <b>(R239C)</b>	21	2		23	Brenner et al 2001 Rodriguez et al 2001 Shiroma et al 2001 Gorospe et al 2002 Shiihara et al 2002 Shiroma et al 2003 Brockmann, Meins et al 2003 Wakabayashi et al 2005 Li et al 2005 van der Knaap et al 2005 Li et al 2006
4	716G→A <b>(R239H)</b>	6	1		7	Brenner et al 2001 Rodriguez et al 2001 Li et al 2005
4	716G→C <b>(R239P)</b>	4			4	Meins et al 2002 Brockmann, Meins et al 2003 Li et al 2005 van der Knaap et al 2005

Exon	Nucleotide Change (amino acid change)	Infantile	Juvenile	Adult	Total Affected Individuals	References
4	724T→G (Y242D)	1			1	Gorospe et al 2002
4	731C→T (A244V)	1	1		2	Aoki et al 2001 Li et al 2005 van der Knaap et al 2005
4	758C→G (A253G)	1			1	Li et al 2005 van der Knaap et al 2005
4	773G→C (R258P)	1			1	Brenner et al 2001
5	827G→T (R276L)			2 <sup>7</sup>	2	Namekawa et al 2002
5	835A→G (K279E)		1		1	Li et al 2005
6	992T→C (L331P)	3 <sup>8</sup>			3	Shiihara et al 2004
6	(349HLins)	1			1	Li et al 2005
6	1055T→C (L352P)	2			2	Bassuk et al 2003 Li et al 2005
6	1073C→T (A358V)	1			1	Dinopoulos et al in press
6	1074C→G (L359V)		1		1	Li et al 2005
6	1086→C (E362D)		1		1	Sawaishi et al 2002
6	1090G→C (A364P)	1			1	Li et al 2005
6	1096T→C (Y366H)	1			1	Li et al 2005
6	1111G→C (E371G)	1			1	Kawai et al 2006
6	1117G→A (E373K)	3			3	Gorospe et al 2002 Li et al 2005
6	1117G→C (E373Q)	1			1	Li et al 2005
6	1121A→G (E374G)	1			1	Li et al 2005
8	1246C→T (R416W)	2	4	3	9	Brenner et al 2001 Gorospe et al 2002 Kinoshita et al 2003 Thyagarajan et al 2004 <sup>9</sup> Li et al 2005 van der Knaap et al 2006 <sup>10</sup>
	<b>TOTAL CASES</b>	86	33	18	137	

1. All six individuals belong to a three-generation family; five are symptomatic, one asymptomatic [Stumpf et al 2003].
2. Two individuals form a pair of monozygotic twins [Meins et al 2002].
3. All three affected adults belong to the same pedigree [Okamoto et al 2002].
4. The individual reported in this manuscript is the same as Patient 10 in Gorospe et al 2002.
5. Asymptomatic mother of affected patient also tested positive for the mutation [Brockmann, Dechant et al 2003].
6. Two patients are identical twins [Li et al 2005].
7. Two individuals are brothers [Namekawa et al 2002].
8. All three patients belong to the same family. The proband showed megalencephaly at 4m with no other abnormalities. MRI at 16m revealed abnormalities in the frontal white matter and basal ganglia. An elder sister and the mother showed the same base change but were asymptomatic and showed only mild abnormalities in the frontal and caudate regions by MRI [Shiihara et al 2004].
9. Two are mother-son [Thyagarajan et al 2004].
10. Two are mother-son [van der Knaap et al 2006].

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